Speech-Language Disorder 1: **FOXP2** Gene Deletion/Duplication

**Test Code:** DFOXP  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

**Condition Description**

Mutations of the **FOXP2** gene (7q31) cause a severe form of speech and language disorder. In a large multi-generational family, a heterozygous missense mutation segregates with family members who have verbal dyspraxia or a deficit in sequencing the complex coordinated orofacial movements required for speech. It is also accompanied by a wide range of linguistic and grammatical deficits. Additionally, the **FOXP2** gene was found to be disrupted by a de novo balanced translocation in a patient with speech and language disorder.

**FOXP2** encodes a transcription factor which likely regulates gene expression in the developing lung, cardiovascular, intestinal, and neural tissue.

**References:**
- OMIM #605317: **FOXP2** gene
- OMIM #602081: Speech-Language Disorder 1

**Genes**

**FOXP2**

**Indications**

This test is indicated for:
- Confirmation of a clinical diagnosis of speech-language disorder 1 in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of speech-language disorder 1 in whom sequence analysis was negative.

**Methodology**

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

**Specimen Requirements**

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:  
Infants (2 years): 3-5 ml  
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

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Special Instructions

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

Related Tests

- Sequence analysis of the $FOXP2$ gene is available and is required before deletion/duplication analysis.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.
- X-Linked Intellectual Disability panels are available for 30, 60, and 90+ genes.